

**Illinois Department of Public Health
Genetic and Metabolic Disease Advisory Committee (GMDAC)
Meeting Minutes – May 21, 2019**

Holiday Inn Chicago SW – Countryside Conference Center
Countryside, IL

Members Present:

Carrie Balian, EHDI parent
Barbara Burton, M.D., Lurie Children's Hospital
Glynis Cailteux, R.N., Kankakee County Health Department
Janine Kahn, M.D., Lurie Children's Hospital
Rachel Katz, L.S.W., Lurie Children's Hospital
George Hoganson, M.D., University of Illinois at Chicago
Beth Leeth, M.S., Lurie Children's Hospital
Melanie Makhija, M.D., Lurie Children's Hospital
Shannon O'Brien, R.D., University of Illinois Medical Center
Erin Paquette, M.D., J.D., Lurie Children's Hospital

Other Attendees:

Rich Dineen, C.G.C., Rush University
Lauren Hitchens, A.P.N, Lurie Children's Hospital
Talana Hughes, M.S., Sickle Cell Disease Association of Illinois
Tess Rhodes, R.N., Division of Specialized Care for Children
Akira Yoshi, M.D., University of Illinois at Chicago
Beth, Jared, Celia and Benny Robinson-GAMT affected family
Emily and Caroline Sans-family of a child with GAMT
Mathew and Emily Partner-parents of child with GAMT

IDPH Representatives:

Matt Charles, Chief Division of Laboratories
Vineet Dhiman, Ph.D., Molecular Section
Yen Ping, Ph.D., Tandem Mass Spectrometry Section
Khaja Basheerrudin, Ph.D., Tandem Mass Spectrometry Section Supervisor
Pam Smith, Newborn Screening Program Manager
Shannon Harrison, Newborn Screen Follow-Up Nursing Supervisor
Laura Ashbaugh, Newborn Screening Follow-Up Nursing Supervisor
Chrissy Knepler, Newborn Screening Follow-up Nursing Supervisor
Ginger Mullin, Newborn Hearing Screen Program Coordinator
Rebecca Barnett, Grants Coordinator

Welcome and Introductions:

Meeting was called to order at 10:32 A.M., and Pam Smith welcomed attendees. Pam Smith stated she is the new Newborn Screening Follow-up Program Manager. Attendee introductions were made. Pam stated that at the current time, there are no vacancies on the committee.

Review of Bylaws, Committee Members, Terms and Vacancies:

Dr. Burton asked the committee to review the changes made to the bylaws. She stated they are not ready for a vote; the program is asking for feedback from the committee at this time. Pam highlighted the following areas: changing the name of the committee to the “Universal Newborn Screening Advisory

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Committee,” including the EHDI program in the bylaws, increasing the committee member appointments to four years, changing the chair’s term appointment, adding an EHDI parent to the committee, and including Early Intervention on the committee. Suggestions were made to add a geneticist and a pediatric neurologist to the list of recommended members. Pam asked that members send any additional input to her.

Approval of Minutes-May 24, 2018 Meeting:

A motion to approve the May 24, 2018 meeting minutes was made by Dr. Makhija. Dr. Khan seconded the motion. All committee members were in favor. Meeting minutes were approved.

IDPH Reports:

Newborn Screening Laboratory

Matt Charles outlined the IDPH laboratory policy for residual newborn screening blood spots. He stated the current retention time is four months for all normal specimens, after which time they are destroyed. The retention time is six years for positive specimens, after which time they are destroyed. The laboratory has received requests to release specimens for research purposes in the past, but Illinois rules do not allow for this. Therefore, a statutory change would be needed in order to release blood spots. Screening by Mayo Clinic Laboratories for Krabbe disease is the only exception to releasing specimens to outside entities since Mayo is performing additional tiers of testing.

Dr. Burton stated she would like an administrative rule change to release specimens with consent from the patient’s family. Dr. Khan agreed and stated the consent could be for the family to retrieve the specimen, so IDPH would not have to send it anywhere else or perform additional tests. No objections were noted from the committee. Matt Charles asked Dr. Burton to send him proposed language for a consent to release the specimen to parents for clinical purposes only and not for research. Dr. Hoganson asked the laboratory to look at keeping the dried blood spots available for a longer period of time.

Dr. Burton stated she has been asked by neonatologists why NICU patients are supposed to have a specimen collection when they are less than 24 hours newborn screen of age. IDPH staff stated this recommendation was made by the Clinical and Laboratories Standards Institute and was a national recommendation for states to follow. The NICU protocol was discussed as well as future changes that will be made regarding reporting of these results to the committee.

Matt Charles and Yen Ping gave a PowerPoint presentation on the progress of adrenoleukodystrophy (ALD) screening. The projected start date is June 3, 2019 with two pilot hospitals and statewide screening in mid-June.

Vineet Dhiman gave a PowerPoint presentation on cystic fibrosis screening. IDPH switched to the Agena IPLEX Pro Panel on March 1, 2018. He stated that although the Agena panel takes longer to run the screen, the Agena panel screens has more mutations.

Vineet Dhiman also presented on the status of spinal muscular atrophy (SMA) screening. He stated the plan is to screen only for the SMN 1 gene and multiplex SMA screening into the existing SCID PCR assay. The lab is currently testing on de-identified dried blood spots to determine if screening can work on high throughput. The target start date is June of 2020. There was discussion regarding the possibility and feasibility of reporting carrier status. IDPH will convene a work group to discuss these types of matters before they implement SMA screening.

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Matt Charles stated the Program has staff hired to work a Tuesday-Saturday schedule. The lab is doing a phased approach: hiring, completing Friday specimen runs on Saturdays, receiving and accessioning specimens on Saturdays, then initiating testing for specimens received on Saturdays.

Matt Charles stated the Program had their NewSTEPs APHL site visit in April of 2018. The committee pointed out Program strengths and weaknesses and outlined recommendations for improvement. The Program has already initiated a number of quality improvements that were identified.

Newborn Screening Follow-Up Program

Pam Smith stated effective January 1, 2019, the newborn screening fee was increased to \$128 per specimen for the implementation of SMA screening. Carrie Balian asked if there is a way to increase the fee to support the EHDI Program as well as to screen for CMV since currently none of the screening fees are going to the EHDI Program. DSAC services and follow-up of abnormal hearing results were discussed.

Pam Smith stated the Follow-up Program currently has a nurse and three support staff working a Tuesday-Saturday schedule. The biggest issue they have encountered is contacting physicians on Saturdays.

Shannon Harrison stated the Program recently implemented the UPS web portal “CampusShip” with all birthing facilities who currently ship their specimens using UPS. CampusShip is a quality improvement initiative that allows for streamlined access to produce shipping labels as hospitals need them. It also includes several user-friendly aspects such as automatic email delivery alerts, timely status alerts of where all shipments are in the pipeline, and integrated ordering of shipping supplies. These services also enable UPS to be more responsive in addressing shipments that are delayed in transit for any reason. In April, IDPH and UPS hosted two web-based training sessions to cover various aspects of CampusShip. Full implementation of CampusShip is expected to begin on June 1, 2019.

Pam Smith and Laura Ashbaugh discussed ALD implementation. The Program will refer patients with abnormal screening results to biochemical geneticists. If they are diagnosed with disease, the geneticists will refer the patient to the appropriate specialists. There was discussion regarding referring patients to medical geneticists and criteria for being an IDPH consultant for ALD.

Pam Smith and Shannon Harrison discussed SMA implementation. Several questions arose regarding if patients could start with a referral to genetics, then be referred to pediatric neurology if diagnosed with SMA. The committee agreed a work group should be formed to discuss such items.

Pam Smith and Matt Charles discussed the status of GAMT legislation that has been proposed. It passed in the House of Representatives and is currently moving through the Senate.

Rebecca Barnett reported that the IDPH grants were delayed due to the new Grants Accountability Transparency Act (GATA) requirements. The approximate funding for fiscal year 2019 is 2.7 million dollars for genetic counseling services, local public health department case finding and referral and pediatric hematology follow-up for sickle cell disease. There are 16 genetic counseling grants at 1.7 million dollars. There are 33 local health departments who participated in \$716,000 grant funds. Eight

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sickle cell clinics were awarded \$306,000. July 1, 2019 is the start date for the 2020 fiscal year applications.

Early Hearing Detection and Intervention Program (EHDI)

Ginger Mullin reported on the EHDI program which works towards the national 1-3-6 goals for hearing screening and follow-up. Recent accomplishments include: integration with the Illinois birth certificate system; updating the list of pediatric audiology providers; completion of the Centers for Disease Control and Prevention annual data survey; and state office implementation of HiTRACK 5 (the new EHDI-information system platform). Ginger reported the current federal initiatives include increasing the use of data exchange; improving timeliness of screening/diagnosis/intervention; reducing loss to follow-up and improving the state's data information system. Finally, programmatic data was shared revealing: statewide greater than 98% of infants are screened with an overall refer rate of 3.2%; current incidence is 2.1/1000; and the loss-to follow-up for the 2017 birth cohort was 30% down from 78% for the 2013 birth cohort.

Other Reports:

Newborn Screening and Laboratory Subcommittee

The Laboratory Subcommittee met for the first time in two years via teleconference on January 23, 2019. Several areas were discussed including <24-hour specimen collection for NICU patients, the need to standardize age appropriate cutoffs, the high false positive rate for galactosemia, and a plan to work on these issues while the laboratory director is taking a leave of absence.

Newborn Screening Cystic Fibrosis Collaborative

There has been no recent meetings or activity.

Newborn Screening Hemoglobinopathy Collaborative

Shannon Harrison reported the Hemoglobinopathy Collaborative continues to meet every other month by teleconference. There are currently 11 hematology centers as well as 2 centers in St Louis on the calls along with SCDAI and STORM. The collaborative discusses QI projects, sickle cell disease events, and issues with testing, insurance, referrals, medication coverage, etc.

Severe Combined Immune Deficiency (SCID) Newborn Screening Collaborative

There has been no recent meetings or activity.

Newborn Screening Expansion Subcommittee

There has been no recent meetings or activity.

Reports from Partners:

Center for Jewish Genetics

Jason Rothstein was not present, and no update was given.

Sickle Cell Disease Association of Illinois (SCDAI)

Talana Hughes reported that SCDAI is focusing on partnerships with health departments to help them reach as many patients as possible. They assist newly diagnosed sickle cell disease patients as well as teenage patients while transitioning to adult care. They have been focusing on services for patients and making connections between patients, partners and stakeholders.

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Illinois Society of Genetic Professionals

Lauren Hitchens stated the organization changed their name to Illinois Society of Genetic Professionals.

Sudden Infant Death Services of Illinois, Inc.

Nancy Maruyama was not present but sent a report to IDPH. IDPH provides grant funding for safe sleep education and bereavement services for deaths on children from birth to age one. The cause of death is no longer called SIDS, but “death by unsafe sleep situations.” As part of NICHQ, SIDS conducted a survey on Safe Sleep practices from birth to age one year. The survey was given to birthing hospitals with survey questions such as “What are your sleep policies?” and “Do you have a sleep policy?” SIDS of Illinois is working with UIC epidemiology to interpret the data. The Illinois SIDS rate is 6 per 1000 with 50% of deaths in Cook County. SIDS of Illinois was formed in 1968 and is celebrating their 50-year anniversary.

Division of Specialized Care for Children

Tess Rhodes from the Division of Specialized Care for Children (DSCC) reported that DSCC has 11 regional offices throughout Illinois, provides care for children age’s birth to 21 years old and covers diagnostic testing for many disorders included in the newborn screening panel not covered by insurance. Their goal is partner with patients and providers to make necessary referrals. Their new assessment is family focused for ongoing care coordination. DSCC has a specific form for anyone doing diagnostic testing to submit to DSCC to start a referral and will contact the family even if the patient doesn’t qualify. They also offer care coordination and financial assistance for medical plans, transportation, and out of state care. She encouraged to the group to partner together since DSCC now covers costs that weren’t previously covered, such as sickle cell disease. ALD and SMA will also be covered. Tess will finalize their new referral form and post it on their website. Rich Dineen requested that the county finder be reinstated on their website, so centers know which office they need to refer their children to.

Other New Business:

Members stated that the group needs to look at the subcommittees and determine if they are still functional.

Public Comments:

Several parents attended the meeting to discuss screening for Guanidinoacetate Methyltransferase (GAMT) Deficiency. An overview was given, and parents asked if Illinois was planning on initiating screening for it. Matt Charles stated he has been talking with the Michigan NBS Program who is trying to implement GAMT screening into their existing Neobase assay. He also outlined the costs and a timeline if Illinois were to implement screening.

Next Meeting Date:

The date for the next meeting was not established, but a late fall 2019 meeting via videoconference was proposed. The group also discussed having all meetings in Chicago. The group indicated that in-person meeting were the preference of the majority. IDPH will send out a Doodle Poll for member availability to schedule a fall meeting. The meeting was adjourned at 1:35 PM.